METABOLIC DISEASE

ORGANIZATIONAL GUIDELINES

Organizational Guidelines represent the minimum requirements for providing care for individuals with metabolic disease. Care and treatment should be provided in a manner with includes adherence to and consistence with each of the following Guidelines.

CRS Enrollment:

All members with metabolic disease diagnoses must be enrolled in metabolic clinics meeting these Guidelines.

Interdisciplinary Team Membership:

Interdisciplinary Team Members must be present during regional clinics and team conferences to review the patient information and determine the need to see the patient at a clinic site and must be available for inpatient consultation or coordination of care with inpatient staff.

Two Interdisciplinary Team Guidelines are identified, one for those conditions which are amenable to nutritional management and one for conditions which are not amenable to nutritional management. The following Team Members must attend outpatient clinics and team conferences and must be available for inpatient consultation or coordination of care with inpatient staff:

Conditions which are amenable to nutritional management:

- Lead Physician - Geneticist with expertise in metabolic disorders, preferably a biochemical geneticist.
- Genetics Counselor or Genetics Nurse
- Metabolic Nutritionist
- Child Psychologist (available)
- Registered Nurse Coordinator (may be the same as the Genetics Nurse)
- CRS Member / Caregiver
- Primary Care Physician[1] (invited)

Conditions not currently amenable to nutritional management:

- Lead Physician - Geneticist
- Genetic Counselor or Genetics Nurse

[1] The Primary Care Physician will be invited to all Team meetings; however, it is understood that PCP will not always be able to attend.

- CRS Member / Caregiver
- Registered Nurse Coordinator (may be the same as the Genetics Nurse)

Available Personnel:

The following personnel must be available during the specialty clinics as needed to see the patient:

- Advocate
- Child Life Specialist
- Educators (education advocacy)
- Social Worker
- Translator

Consultative Personnel:

Consultation may be obtained as needed for an eligible patient when the consultation is related to the diagnosis or regarding a potential condition that is CRS eligible. The list of disciplines which the CRS Clinic must have access to for consultation includes but is not limited to the following:

- Pediatric Neurologist
- Orthopedist
- Neurosurgeon
- Ophthalmologist
- Physical Therapist
- Nutritionist
- Child Psychologist
- Cardiologist
- ENT
- Physical Therapist
- Occupational Therapist
- Pediatrician
- Audiologist

Outreach Clinics:

Outreach clinics are designed to provide a limited specific set of services including evaluation, monitoring, and treatment in settings closer to the family than a regional clinic. Major treatment plan changes must be communicated to the regional clinic.

Members with metabolic conditions may attend neurology, genetics, or other Outreach Clinics as determined by the Interdisciplinary Team.

Facilities & Services:

1. Age-appropriate settings for all patients
2. Defined age-appropriate services, i.e., Pediatrics, Adolescent Medicine, and/or Internal Medicine
3. Pediatric and Adult Intensive Care Units
4. Nutrition or Dietary Department
5. Social Work Department
6. Identified clinic area
7. Laboratories:
    a) performing pediatric phlebotomy
    b) performing routine pediatric lab evaluations (if contracted, the contracts must address quality and time requirements)
    c) performing or providing specialized genetics testing relevant to the condition
8. Access to the pharmacy and/or a distribution point for metabolic formula.

Team/Staff Meetings:

Team and staff meetings will be held based on the age of the patient and their diagnosis. At a minimum the following will occur:

1. Interdisciplinary Team Meetings - patient specific meetings held with the family for review of the status and planning the course of treatment:
    a) At the time of initial diagnosis and
    b) Once every three years thereafter (at a minimum.)
2. Staff meetings annually to focus on issues of clinic patient care and clinic administration.
3. Education meetings annually to focus on new information regarding the care and treatment for patients with metabolic diseases.

Lead Physician Specialists:

Qualifications: The lead physician for patients with metabolic diseases amenable to nutritional management shall be a Board Eligible/Board Certified in genetics, preferably a biochemical geneticist, with pediatric experience.

The lead physician for patients with metabolic disease not related to dietary conditions/treatment shall preferably be a biochemical geneticist with pediatric experience.

GUIDELINES FOR PATIENT SERVICES, EVALUATION AND MONITORING FOR METABOLIC DISEASE

The purpose of these guidelines is to promote a uniform level of care at CRS Clinics for members with metabolic disease, and to provide a general framework for good patient care. Their relevance to specific situations will depend on individual variations in clinical course and professional judgment. In addition, this document should serve as a tool to assess programs, secure resources needed to enhance patient care and education, and guide the future growth and development of metabolic care.

Diagnosis:

Goal: There is a vast, and ever increasing, number of medically defined inborn errors of metabolism. Diagnosis of specific conditions will be dictated by the biochemical basis for the individual condition. To provide accurate and timely diagnosis of metabolic disease the following guidelines are offered recognizing that diagnosis for specific conditions will be individualized:

Defects of Intermediary Metabolism such as Amino Acid and Organic Acid Disorders - Documentation of inborn error of metabolism, including but not exclusive of abnormalities in amino acid and organic acid metabolism, includes the following information needs:

1. Complete medical history
2. Complete physical examination
3. Copies of laboratory reports documenting the purported inborn error of metabolism, including but not limited to quantitative plasma amino acids and quantitative urinary organic acids

Metabolic Storage Diseases - Documentation of metabolic storage disease requires the following information:

1. Complete medical history
2. Complete physical examination
3. Laboratory data documenting the metabolic storage disease including but not limited to pathologic demonstration of stored material and enzymatic confirmation of a specific defect

Confirmation of the diagnosis requires:

Confirmation of diagnosis by a pediatric geneticist, pediatrician, or pediatric neurologist.

Evaluation and Education of Newly Diagnosed Patients

Goal: To provide accurate assessment of physical and emotional status, and to begin patient and family education.

The following assessments should be completed:

1. Psychological Evaluation including family members and family issues initially and on an ongoing basis as determined by the Interdisciplinary Team.
2. Psychometric Evaluation using nationally accepted instruments appropriate to the age of the patient and family member(s). Baseline and periodic assessments should be completed.

Goal: To help families cope with the emotional impact of diagnosis and formulate an appropriate therapeutic plan.

1. Information should be provided to the family following a positive newborn screen by the Newborn Screening Follow-up Staff if suspected metabolic condition is one identified by the newborn screen. If the metabolic disease is one suspected because of the clinical presentation, information shall be provided as iterated in #2 below.
2. Prior to or concomitant with the confirmation testing, experienced metabolic personnel should provide families with information about the suspected metabolic disease including the diagnosis and treatment.
3. Following confirmation, a geneticist and/or a genetic counselor should provide appropriate genetics counseling to the family.

   If available, written information regarding the condition should be provided, such as diagnostic requirements, inheritance concerns, possible treatment regimens, and prognosis.
4. A nutritionist should provide general information about nutritional management and respond to questions regarding their specific nutritional program.
5. Information about the CRS experience; i.e., what is CRS, who does CRS serve, what is a metabolic clinic like, who will be there, what is the family's role, etc.
6. Information regarding support groups and how to contact them.

Ongoing Patient Evaluation and Monitoring:

Goal: The ongoing patient evaluation and monitoring will be dictated by the biochemical basis for individual condition. However, some generalizations are appropriate to iterate as guidelines for the care of this group of disorders as a whole and to anticipate and treat physical and psychosocial problems and complications of the disease.

Clinic Visits:

1. For disorders of Intermediary Metabolism Amenable to Nutritional Management including but not limited to Amino and Organic Acid Disorders:
   a. Monthly for the first 6 months of life
   b. Bimonthly from 6 months to a year
   c. Quarterly from one year to 3 years

d. Semiannually thereafter

2. For Storage Disorders
   a. Bimonthly for the first 6 months
   b. Quarterly from 6 months to a year of age
   c. Semiannually from one year to 3 years
3. For Conditions Not Amenable to Nutritional Management
   a. Quarterly to one year of age
   b. Annually thereafter

Monitoring:

Metabolic patients are monitored by laboratory analysis including but not limited to metabolic analysis of blood and urine.

Treatment:

Goal: The treatment for inborn errors of metabolism will be determined by the specific condition. However, some generalizations are appropriate to iterate as guidelines for the care of this group of disorders as a whole. Broadly, the inborn errors can be divided into two groups: those amenable to treatment by nutritional management (including inborn errors of vitamin metabolism) and those not amenable. The following are given as guidelines for these two categories recognizing that treatment will be individualized to the specific condition.

The Lead Physician of the Metabolic Team and Team members must be involved in decisions regarding all major procedures.

1. For Conditions Amenable to Nutritional Management (Including Inborn Errors of Vitamin Metabolism):
   a. Provide metabolic formula and foods to limit the intake of the offending metabolite while supporting optimum growth and development.
   b. Provide pharmacologic administration of vitamin(s) to correct or improve the metabolic disruption.
2. For Conditions Not Amenable to Nutritional Management:

   Consequences and complications are managed within the specialty clinics. These actions must be coordinated with the Interdisciplinary Metabolic Clinic Team.

Gastrointestinal System/Nutrition:

Goal: To anticipate and treat nutritional deficits and complications. The ultimate goal is to achieve optimum growth and nutrition. Continued monitoring of growth

parameters in conjunction with appropriate metabolic nutritional guidance is extremely important.